Case Report

Laurence Moon Bardet Biedle Syndrome

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Abstract

Laurence Moon Bardet Biedle syndrome is a rare, autosomal recessive genetic disorder involving multiple systems and has wide spectrum of clinical features. Characteristic features of this disorder are retinitis pigmentosa, polydactyly, truncal obesity and learning difficulties. It may also be associated with hypogonadism in male and complex genitourinary abnormalities in female. We present a case of 33 years male patient having obesity, decreased vision, polydactyly, hypogonadism and retinitis pigmentosa. These clinical features are consistent with Laurence Moon Bardet Biedle syndrome.

Keywords: Polydacytyly, Retinitis Pigmentosa, Brachydactyly, Syndactyly, Hypogonadism, Hypodontia.

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Introduction

Laurence-Moon syndrome is caused by changes (mutations) in the PNPLA6 gene and is inherited in an autosomal recessive manner[1]. PNPLA6 gene encodes neuropathy target esterase, critical in phosphatidylcholine metabolism, membrane phospholipid trafficking and axonal integrity[2].

The incidence is much higher in some populations with a high level of consanguinity of marriage or those that are geographically isolated. The syndrome is more often seen in the Arab population especially in the Bedouin population of Kuwait, affecting about 1 in 13,500 newborns and also on the island of Newfoundland (off the east coast of Canada), where the prevalence is 1 in 17,000 newborns[3,4].

Larger studies in Bangladeshi population are needed and the literature review has revealed only few cases[5].

The Laurence moon bardet biedle is a rare autosomal recessive disorder, which is characterized principally by cardinal symptoms of marked central obesity, retinal dystrophy, polydactyly, mental retardation and hypogonadism and renal dysfunction.

Other features not always present include hepatic fibrosis, diabetes mellitus, neurological, speech and language deficits, behavioral traits, facial dysmorphism, dentalanomalies and developmental delay.

The authors present a case of LMBBS presented with hypogonadism, marked central obesity, retinitis pigmentosa and polydactylyetc along with a brief review of the literature.

Case Report

A 33 years-old-male patient from Fulbarigate, Khulna has presented with the complaints of progressive dimness of vision more marked at night from his childhood. On query he gives history of nocturia, polydipsia, loss of libido, absent penile erection. There is no history of consanguity of marriage; no developmental delay. No other family member has similar problem. He is diabetic for 20 years and is on regular insulin therapy.

On examination his height is 141 cm, weight 58 kg; resulting BMI = 29.2. He has short stature and short neck. There is bilateral gynaecomastia (Fig-1), testicular atrophy and small penis (2 cm).

Figure-1: Gynaecomastia, Obesity. (Source: The reporting case).

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There is polydactyly involving both upper limbs and right lower limb. There is also brachydactyly in right foot and syndactyly of left foot (3rd and 4th toes) (Fig -2 & 3).

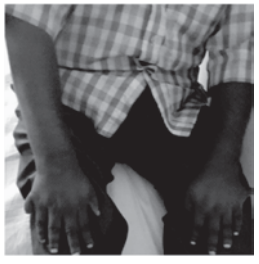
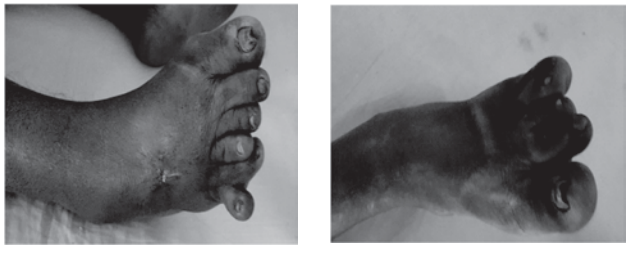


Figure-2: Polydactyly.
(Source: The reporting case).



(a) (b)

Figure-3 :(a & b) Polydactyly, Bachydactyly, Syndactyly.

(Source: The reporting case).

His pulse is 80 /min; BP- 120/70 mm Hg. There is no anaemia, jaundice, cyanosis, clubbing or koilonychia. He has no thyromegaly or lymphadenopathy.

His eyes are small and deep seated. On ophthalmoscopy there is bilateral retinitis pigmentosa. Optic disc, vessels were normal. His fundal photograph was taken and shows bilateral retinitis pigmentosa (Fig-4).

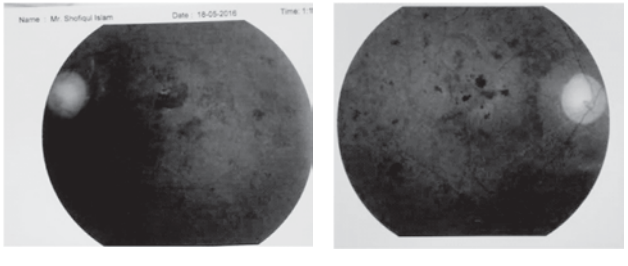


Figure-4: Retinitis Pigmentosa (a) right eye; (b) left eye.

(Source: The reporting case).

Others systemic examinations reveals no abnormality.

His Blood TC DC ESR, urine R/M/E and USG of whole abdomen is normal. X ray chest and ECG reveals normal findings. Blood sugar is normal with insulin therapy. Urine Routine and microscopic examination reveals - normal. USG of testes reveals bilateral small testes. Serum testosterone level is markedly reduced (0.1ng/ml).

Discussion

In 1866 Laurence and Moon described a disease characterized by adiposity, genital dystrophy, retinitis pigmentosa, and mental deficiency, affecting four members of one family. In 1920 Bardet described a similar syndrome in a child with polydactyly. In 1922 Biedl reported a similar condition in a brother and a sister. In 1925 Solis-Cohen and Weiss suggested the name Laurence-Moon-Biedl syndrome for this condition[6].

Five cardinal signs were considered the major components of LMBB syndrome. They were retinitis pigmentosa, mental retardation, polydactyly, syndactyly and obesity.

Along with the characteristic features, the patients with this syndrome at times present with other findings like intellectual impairment, cardiovascular abnormalities, deafness and dental anomalies[7,8].

Our patient was 33 year old. Polydactyl has been reported in upper or lower limbs. Most of the reported cases were females, whereas our patient was male. Our patient hadpolydactyly, brachydactyly, syndactyly in lower limb. In 75 % cases obesity was found with mean BMI to be 33 kg/m^2. Our patient was also obese with BMI 29.2.

As the clinical variability of the syndrome is very large, Klein and Amman have adopted the following classification:

1. Complete form:

2. Incomplete form:

3. Atypical form

4. Extensive form

Modified diagnostic criteria for Bardet-Biedl syndrome[9].

Primary features

- Rod-cone dystrophy
- Polydactyly
- Obesity
- Learning disabilities
- Hypogonadism in males
- Renal anomalies

Secondary features

- Speech disorder/delay
- Strabismus/cataracts/astigmatism
- Brachydactyly/syndactyly
- Developmental delay
- Polyuria/polydipsia (nephrogenic diabetes insipidus)
- Ataxia/poor coordination/imbalance
- Mild spasticity (especially lower limbs)
- Diabetes mellitus
- Dental crowding/ hypodontia/small roots/high arched palate
- Left ventricular hypertrophy/congenital heart disease
- Hepatic fibrosis

For diagnosis of this syndrome the patients should have either four primary features or three primary and two secondary features[10,11,12,13].

Our patient had 4 primary features (Rod-cone dystrophy, Polydactyly, Obesity, hypogonadism) and 2 secondary features (brachydactyly/syndactyly, diabetes mellitus). Hence we diagnosed the patient to be suffering from Laurence Moon Biedl Bardet syndrome.

Initial investigation and follow up recommendations for a person with Bardet-Biedl syndrome[14].

Baseline

- Electroretinogram (ERG)/Visually Evoked Responses (VER)
- Renal ultrasound
- Intravenous pyelogram (IVP) or DMSA/DPTA scan
- ECG & echocardiogram
- Prader-Willi syndrome exclusion by molecular testing

Consider

- CT/ MRI of brain
- Electroencephalogram (EEG)
- Statementioning of educational needs
- Registration of blindness
- Speech assessment & therapy

Six monthly

- Urine analysis (dipstick)

Conclusion

It has got poor prognosis. Quality of life and survival of patient depends on these verities of the condition and treatment provided.

Acknowledgement

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2. Eye and Laser Vision Hospital, Khulna.

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